



AMHR2 gene

anti-Müllerian hormone receptor type 2

Normal Function

The *AMHR2* gene provides instructions for making the anti-Müllerian hormone (AMH) receptor type 2, which is involved in male sex differentiation. The AMH receptor type 2 is found on the surface of Müllerian duct cells. The Müllerian duct, found in both male and female fetuses, is the precursor to the female reproductive organs. During development of male fetuses, cells of the testes release a protein called the AMH protein. The AMH protein attaches (binds) to the AMH receptor type 2, which signals self-destruction (apoptosis) of the Müllerian duct cells. As a result, the Müllerian duct breaks down (regresses) in males. In females, who do not produce the AMH protein during fetal development, the Müllerian duct becomes the uterus and fallopian tubes.

Health Conditions Related to Genetic Changes

persistent Müllerian duct syndrome

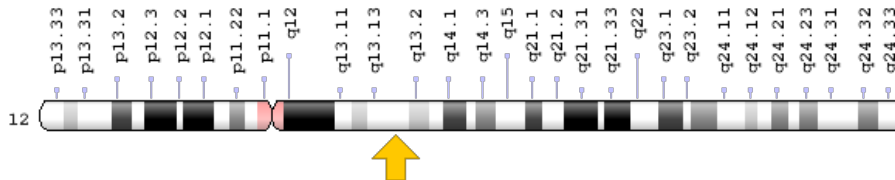
Persistent Müllerian duct syndrome type 2, a disorder of sexual development that affects males, is caused by mutations in the *AMHR2* gene. Males with this condition have female reproductive organs in addition to normal male reproductive organs. At least 24 mutations in the *AMHR2* gene have been identified in people with persistent Müllerian duct syndrome type 2. Most mutations change single protein building blocks (amino acids) in the AMH receptor type 2 protein. Other mutations result in a premature stop signal that leads to an abnormally short protein. Still other mutations delete regions of DNA from the *AMHR2* gene, which changes the instructions for the protein. The most common mutation, a deletion of 27 DNA building blocks (nucleotides), occurs in about half of affected individuals with an *AMHR2* gene mutation.

Mutations in the *AMHR2* gene lead to an abnormal protein that is stuck inside the Müllerian duct cells and not found on the surface. Therefore, the cells cannot receive the signal for apoptosis. As a result, the Müllerian duct persists and becomes a uterus and fallopian tubes. Because the AMH receptor type 2 is not involved in the formation of male reproductive organs, affected males also have male reproductive organs.

Chromosomal Location

Cytogenetic Location: 12q13.13, which is the long (q) arm of chromosome 12 at position 13.13

Molecular Location: base pairs 53,423,855 to 53,431,672 on chromosome 12 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- AMH type II receptor
- AMHR
- AMHR2_HUMAN
- anti-Muellerian hormone type-2 receptor
- anti-Muellerian hormone type II receptor
- anti-Mullerian hormone receptor type II
- anti-Mullerian hormone receptor, type II
- MIS type II receptor
- MISR2
- MISRII
- MRII
- Mullerian inhibiting substance type II receptor

Additional Information & Resources

Educational Resources

- An Introduction to Genetic Analysis (seventh edition, 2000): Development of the Urogenital System
<https://www.ncbi.nlm.nih.gov/books/NBK21846/figure/A3724/>
- StemBook (2008): Müllerian Duct Formation and Neonatal Development
<https://www.ncbi.nlm.nih.gov/books/NBK27042/#uterinestemcells.sec2-1>

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28AMHR2%5BTIAB%5D%29+OR+%28%28AMH+type+II+receptor%5BTIAB%5D%29+OR+%28AMHR%5BTIAB%5D%29+OR+%28MIS+type+II+receptor%5BTIAB%5D%29+OR+%28MISR2%5BTIAB%5D%29+OR+%28MISR2%5BTIAB%5D%29+OR+%28MISRII%5BTIAB%5D%29+OR+%28MRII%5BTIAB%5D%29+OR+%28Mullerian+inhibiting+substance+type+II+receptor%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D>

OMIM

- ANTI-MULLERIAN HORMONE TYPE II RECEPTOR
<http://omim.org/entry/600956>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_AMHR2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=AMHR2%5Bgene%5D>
- HGNC Gene Family: Type 2 receptor serine/threonine kinases
<http://www.genenames.org/cgi-bin/genefamilies/set/346>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=465
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/269>
- UniProt
<http://www.uniprot.org/uniprot/Q16671>

Sources for This Summary

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